Arizona Department of Health Services

Meeting Notice

Newborn Screening Advisory Committee
Meeting Notice and Agenda
Wednesday, December 9, 2015

Pursuant to A.R.S. § 38-431.02, notice is hereby given to the members of the Newborn Screening Advisory Committee (NBSAC) of the Arizona Department of Health Services and to the general public that the NBSAC will hold a meeting open to the public on December 9, 2015 from 12:00 p.m. until 2:00 p.m., at the Arizona State Laboratory, 250 North 17th Avenue, First Floor Igloo Conference Room.

The agenda for the meeting is as follows:

I. Call to Order, Welcome and Introductions (Director Cara Christ, M.D., M.S.)
II. Severe Combined Immune Deficiencies (SCID) Update (Ward Jacox)
III. Panel Addition Proposals – Discussion
   a. Secretary's Advisory Committee on Heritable Disorders in Newborns and Children Additions and Candidate Disorders
   b. Secondary MSMS Disorders
IV. Call to Public
   This is the time for the public to comment. Members of the Committee may not discuss items that are not on the agenda. Therefore, action taken as a result of public comment will be limited to directing staff to study the matter or schedule it for further consideration/decision at a later date.
V. Announcements (Ward Jacox)
VI. Adjournment

A copy of the agenda background material provided to Committee members will be available for public inspection on the AZ NBS website – www.aznewborn.com.

Persons with a disability may request a reasonable accommodation, such as a sign language interpreter, by contacting Ward Jacox at (602) 364-1409 or toll free at (800) 548-8381 (For the hearing/speech impaired, please call 711 for the AZ Relay Service) Requests should be made as soon as possible to allow time to arrange the accommodation.

Dated this 1st day of December, 2015.

ARIZONA DEPARTMENT OF HEALTH SERVICES

Ward B. Jacox
Assistant Bureau Chief
Chief of the Office of Newborn Screening (Acting)

NewSTEPs

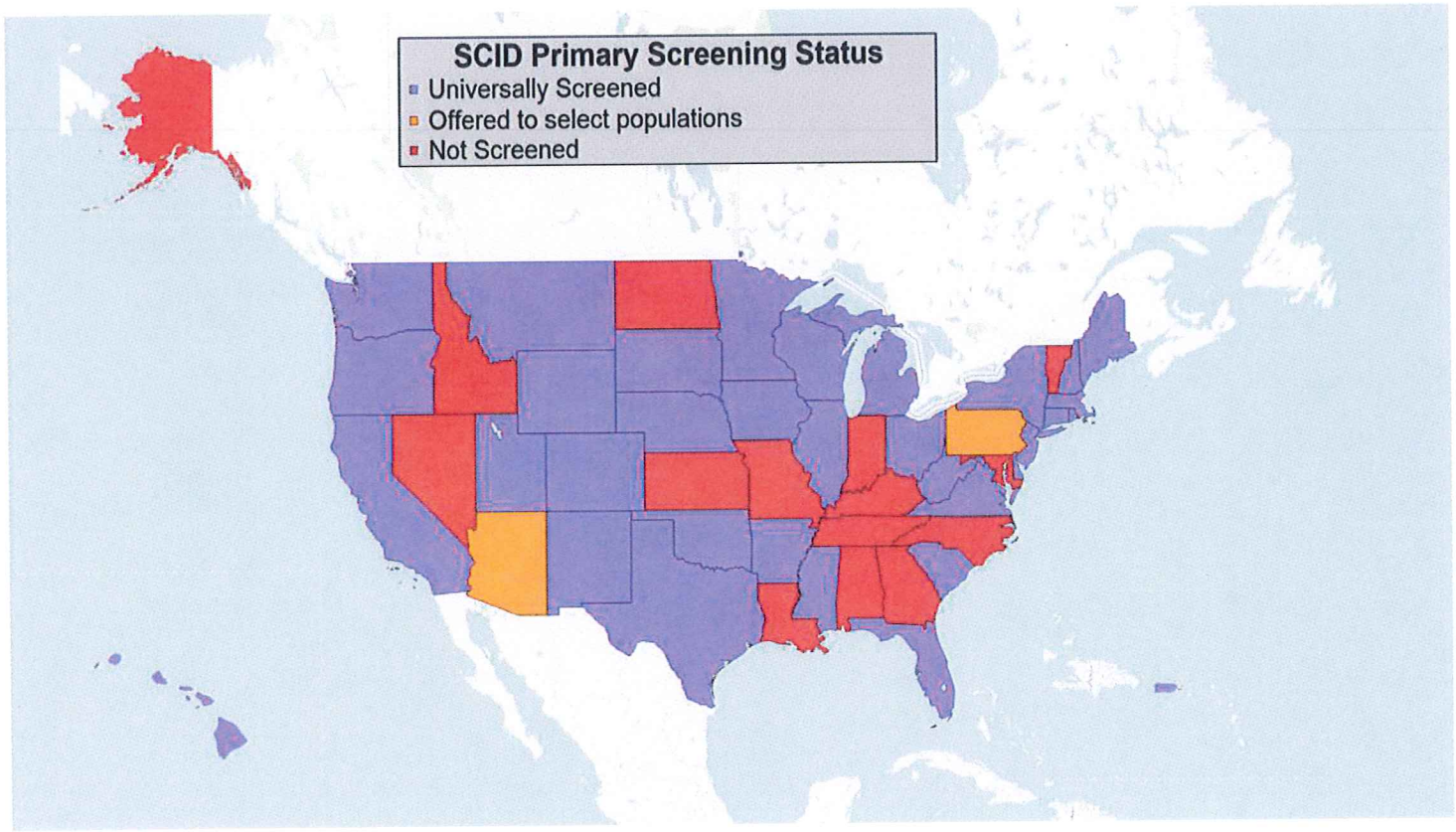
A Program of the Association of Public Health Laboratories™

Severe Combined Immunodeficiencies - SCID
Screening Status by State Newborn Screening Program

SCID Screening Status Report
As of December 4, 2015

The purpose of this document is to report on the Screening Status of SCID in each US Newborn Screening Program. The report includes all 50 states plus Washington D.C. and Puerto Rico.

To update the screening status of SCID, please contact your NewSTEPs State Administrator

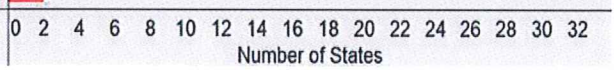


NewSTEPs
A Program of the Association of Public Health Laboratories™

Severe Combined Immunodeficiencies - SCID

SCID Newborn Screening Status Chart

Primary Screening Status	Secondary Screening Status	Tertiary Screening Status	Summary Count
Universally Screened	Required by law or rule	N/A	31
	Not required by law or rule	N/A	4
Offered to select populations	N/A	N/A	2
Not Screened	Pursuing implementation	Performing validation/pilot studies	1
		Required but not yet fully implemented	4
		Seeking authorization/funding	8
	Data Not Provided	Data Not Provided	2



GOALS TABLE - Arizona:

GOAL 1: Legislative Approval to Modify Fee (Second Legislative Session)					
	Activity	Outcome	Start	End	Who is lead?
Step 1	Convene Stakeholder group to discuss SCID status and fee statute	A decision of whether outside stakeholders will pursue a legislative change	N/A	11/23/2015	NBS OC Educator
Step 2	General SCID Outreach and Education (with invited experts)	General public, providers and NBSAC provided with information on SCID and SCID screening	12/2015	6/1/2016	Educator NBS OC
Step 3	Statutory support, if requested by Legislature (with invited experts)	Statute Modification to allow for fee increase	1/1/2016	4/15/2016	NBS OC Educator LL
Note: All subsequent steps will only proceed if legislation to modify the NBS fee succeeds					
Alternative 1: Legislation specifies exempt rulemaking (streamlined process with postponed EIS)					
Step 4	Rulemaking adding SCID (exempt)	Final rule submitted to Secretary of State's Office	4/15/2016	6/1/2016	NBS OC
Step 5	Stakeholder Coordination (with invited experts)	Key stakeholders provided with information on SCID benefits and costs	4/2016	6/1/2016	Educator NBS OC
Step 6	Economic Impact Statement	GRRC approval on rule continuation	6/1/2016	4/2018 (est)	NBS OC
Alternative 2: Legislation approved with regular rulemaking (estimated duration – 1 year)					
Step 4	Rulemaking adding SCID (exempt)	Final rule submitted to Secretary of State's Office	4/15/2016	6/1/2017	NBS OC
Step 5	Stakeholder Coordination (with invited experts)	Key stakeholders provided with information on SCID benefits and costs	4/15/2015	6/1/2017	NBS OC
Step 6	Economic Impact Statement	GRRC approval on rule continuation	TBD	3/2017 (est)	NBS OC
Note: All subsequent steps are based on grant of exempt rulemaking					

NBS: Newborn Screening
OC: Office Chief
LL: Legislative Liaison
SG: Specimen Gate (Perkin Elmer product)
FU: Follow-up

GOAL 2: Infrastructure Selection - Testing					
	Activity	Outcome	Start	End	Who is lead?
Step 1	Evaluate equipment/reagent alternatives	Select instrument platform and reagent source	12/2015	3/2016	NBS Lab
Step 2	Evaluate testing space alternatives (if in-house)	SCID testing space selected	12/2015	3/2016	NBS OC
Step 4	Procure instrument/validation reagents	Instruments and validation reagents in house.	4/15/2016	6/1/2016	NBS Lab
Step 5	SOP development (Lab and F/U)	SOP approved by Laboratory Director	4/2016	6/2016	NBS Lab
Step 6	SG/Neometrics data module development	SCID testing and reporting integrated into data management system.	6/2016	7/2016	NBS Lab NBS FU
Step 7	SCID Validation Study	Validated method and cutoffs	6/2016	8/2016	NBS Lab
Step 8	Complete data management protocol testing	Lab/Follow-up data management modules in production (Go Live)	8/2015	8/2016	NBS Lab NBS FU
GOAL 3: Infrastructure Development – Follow-up/Education					
	Activity	Outcome	Start	End	Who is lead?
Step 1	Convene ad hoc committee to develop follow-up protocols	SCID Follow-up protocol approved by Laboratory Director	4/2016	6/2016	NBS FU
Step 2	Educational materials evaluation, development and approval	Education and outreach materials – final versions for multiple audiences (Eng/Sp/Navajo)	4/2016	7/2016	Educator
Step 3	Integrate SCID outreach into NBS education plan	NBS Education plan version update	4/2016	7/2016	Educator
GOAL 5: SCID Implementation - August 2016					
	Activity	Outcome	Start	End	Who is lead?
Step 1	Implementation outreach	Notification of all relevant stakeholders of SCID Go Live date	6/2016	8/2016	NBS OC Educator
Step 2	SCID Implementation	SCID testing offered to all AZ newborns	8/2016	N/A	NBS OC
Step 3	Draft grant report with lessons learned	Final grant report	6/2016	9/2016	NBS OC

Arizona Department of Health Services

Office of Newborn Screening

Newborn Screening Advisory Committee

September 24, 2014

SCID Estimates for Arizona (~$10/screen)

Population[1]	Incidence	Projected 1st screens	Expected Cases Cases/year	Years to 1st case	AZ FY2016 $10.14 SCID Revenue/Year[6]	Cost to 1st Case[7]
General (not including below)	1/100,000	48,203	0.48	2.07	$488,786	$1,014,021
Hispanic[2]	1/25,000	33,254	1.33	0.75	$337,202	$253,505
AIAN (non-Athabascan)[3]	1/100,000	2,396	0.02	41.74	$24,293	$1,014,021
AIAN Athabascan (Off reservation births)[4]	1/2,000	2,261	1.13	0.88	$22,924	$20,280
Total:		86,113	2.97	0.34	$873,204	$294,357
AIAN Athabascan (on-reservation)[5]	1/2,000	1,328	0.66	1.51	$13,468	N/A

1. Population categories based on mother's reported race/ethnicity.
2. Hispanic incidence is a rough estimate based on early California pilot study data.
3. American Indian Alaska Native (AIAN) (non-Athabascan) incidence is a very conservative estimate, but more likely closer to Hispanic than General.
4. Off reservation birth percentage was estimated from births at non-IHS facilities, then applied to Athabascans.
5. Calculations assume all on-reservation Athabascan births sent out of state.
6. Revenue assumes cost per newborn billed to first screen.
7. Cost based on average annual cost over five years: $873,204

KEY POINTS

- Washington State: "The model predicts a benefit/cost ratio of 4.93, meaning that for every dollar of costs to screen newborns for SCID, there will be almost $5 worth of benefits. Their value of one life saved is estimated at $ 7.7 million. (the last baby born with SCID in California prior to starting screening generated more than $4 million in medical bills - Puck 2012)."
- Washington State's assumed incidence was 1:49,827, which is less than Arizona's combined incidence, due to the contribution of Athabascan AIANs.
- Perkin Elmer's SCID testing kit is expected to be approved by FDA in early 2015. Current ***program*** costs are estimated at ~$10 per screen in-house, while sendout to Perkin Elmer is ~ $6.50 per screen (which does not include follow-up, education, billing and sample handling).

DEPARTMENT OF HEALTH & HUMAN SERVICES

Office of the Secretary
Washington, D.C. 20201

NOV 12 2015

Joseph A. Bocchini, Jr., M.D.
Chairperson
Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

Thank you for your recent letter to Secretary Burwell from the Advisory Committee on Heritable Disorders in Newborns and Children (Committee) submitting the Committee's recommendations to add X-linked Adrenoleukodystrophy to the Recommended Uniform Screening Panel and to provide funding to states to implement screening for this condition. The Secretary asked me to respond to you on her behalf.

Please be assured that these recommendations will receive careful review within the Department. You will receive a more detailed response regarding actions on these recommendations within 120 days, as required by the Newborn Screening Saves Live Reauthorization Act of 2014.

Please accept my personal thanks to you and the members on the Committee for all of your valuable work to improve the health of our nation's infants and children.

Sincerely,

Mary K. Wakefield
Acting Deputy Secretary

THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

OCT 2 8 2015

Joseph A. Bocchini, Jr., M.D.
Chairperson
Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane, Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

I am providing you with an update to the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children's (Committee) recommendation on the addition of Mucopolysaccharidosis Type I (MPS I) to the Recommended Uniform Screening Panel.

After consultation with various components of the Department of Health and Human Services, I have referred the Committee's recommendation to the Interagency Coordinating Committee on Newborn and Child Screening (ICC) for additional review and input. I have instructed the ICC to submit its analysis, advice, and recommendation to me.

Thank you again for the Committee's efforts in conducting an informative evidence-based review of implementing newborn screening for MPS I. I appreciate the Committee's valuable work to improve the health of our nation's infants and children.

Sincerely,

Sylvia M. Burwell

Sylvia M. Burwell

THE DEPUTY SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

MAR 16 2015

Joseph A. Bocchini Jr., MD
Chairperson, Discretionary Advisory Committee
on Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18W68
Rockville, Maryland 20857

Dear Dr. Bocchini:

Thank you for your letter regarding the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children's recommendation to facilitate a national dialogue among federal and state stakeholders on the benefits of measuring succinylacetone in dried blood spots to improve the specificity of newborn screening for tyrosinemia type I. The Secretary asked me to respond directly on her behalf. I have reviewed the Committee's recommendation, as well as the report, *Succinylacetone as a Primary Marker to Detect Tyrosinemia Type I in Newborns and Its Measurement by Newborn Screening Programs,* that were enclosed with your letter.

On behalf of the Secretary, I accept the Committee's recommendation. I have asked the Centers for Disease Control and Prevention (CDC) to facilitate a national discussion to address technical and practice issues in measuring succinylacetone for screening newborns. I am also encouraging CDC, working with the Health Resources and Services Administration and others in the Department, to engage key stakeholders to ensure a robust discussion to improve more consistent implementation of screening for this condition. If you would like more information on CDC's plans and activities regarding your recommendation, please contact Dr. Carla Cuthbert at CCuthbert@cdc.gov.

I appreciate the dedication and commitment of the Committee in addressing issues that impact the health of our nation's newborns and children, and we look forward to working together in the future.

Sincerely,

William V. Corr

THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

MAR 0 2 2015

Joseph A. Bocchini, Jr., MD
Committee Chairperson
Discretionary Advisory Committee on Heritable Disorders
in Newborns and Children
Professor and Chairperson
Department of Pediatrics
Louisiana State University
1501 Kings Highway
Shreveport, LA 71130

Dear Dr. Bocchini:

As indicated in the January 27, 2014 letter from Secretary Sebelius, the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) recommendations regarding the addition of Pompe disease to the HHS Recommended Uniform Screening Panel (RUSP) were forwarded to the Interagency Coordinating Committee on Screening in Newborns and Children (ICC) for additional input regarding implementation.

The ICC reviewed the DACHDNC's recommendations as well as evidence from method evaluation studies, information on test quality, national guidance documents, and current state screening activities. In its report to me, the ICC noted challenges associated with the implementation of state newborn screening for Pompe disease including resource limitations for laboratory testing, management of late-onset cases, and increased burden on treatment and follow-up systems. However, the ICC emphasized that over time, adoption of this recommendation will help increase the number of newborns screened and decrease the morbidity and mortality of babies born with this disease.

I would like to commend the DACHDNC on their review and analysis of benefits and harms of newborn screening for Pompe disease and the ICC report that described the capability of state newborn screening programs to offer comprehensive testing and follow-up for the condition. The information from the objective evidence report, *Newborn Screening for Pompe Disease,* was taken into account as I reviewed the ICC's report.

Taking into consideration the information presented in these reports, I accept the DACHDNC recommendation to add Pompe disease to the RUSP. The Affordable Care Act requires that most health plans cover the evidence-informed preventive care and screenings provided for in the comprehensive guidelines supported by Health Resources and Service Administration (HRSA). Because the RUSP is a component of these guidelines, a condition added to the RUSP must be covered. It should be understood that addition of Pompe disease to the RUSP does not constitute a requirement for states to implement screening, only a recommendation. I recognize

the complex issues surrounding newborn screening for Pompe disease and encourage Federal agencies to support states as they build capacity and implement state-wide screening.

I appreciate the DACHDNC's dedication and continued hard work to improve the health of our nation's infants and children.

Sincerely,

[redacted]

Sylvia M. Burwell

Recommended Uniform Screening Panel[1]
SECONDARY[2] CONDITIONS[3]
(As of March 2015)

ACMG Code	Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Cbl C,D	Methylmalonic acidemia with homocystinuria	X				
MAL	Malonic acidemia	X				
IBG	Isobutyrylglycinuria	X				
2MBG	2-Methylbutyrylglycinuria	X				
3MGA	3-Methylglutaconic aciduria	X				
2M3HBA	2-Methyl-3-hydroxybutyric aciduria	X				
SCAD	Short-chain acyl-CoA dehydrogenase deficiency		X			
M/SCHAD	Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency		X			
GA2	Glutaric acidemia type II		X			
MCAT	Medium-chain ketoacyl-CoA thiolase deficiency		X			
DE RED	2,4 Dienoyl-CoA reductase deficiency		X			
CPT IA	Carnitine palmitoyltransferase type I deficiency		X			
CPT II	Carnitine palmitoyltransferase type II deficiency		X			
CACT	Carnitine acylcarnitine translocase deficiency		X			
ARG	Argininemia			X		
CIT II	Citrullinemia, type II			X		
MET	Hypermethioninemia			X		
H-PHE	Benign hyperphenylalaninemia			X		
BIOPT (BS)	Biopterin defect in cofactor biosynthesis			X		
BIOPT (REG)	Biopterin defect in cofactor regeneration			X		
TYR II	Tyrosinemia, type II			X		
TYR III	Tyrosinemia, type III			X		
Var Hb	Various other hemoglobinopathies				X	
GALE	Galactoepimerase deficiency					X
GALK	Galactokinase deficiency					X
	T-cell related lymphocyte deficiencies					X

1. Selection of conditions based upon "Newborn Screening: Towards a Uniform Screening Panel and System." *Genetic Med.* 2006; 8(5) Suppl: S12-S252" as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics.* 2006; 117 (5) Suppl: S308-S314.